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Surveillance for Anencephaly and Spina Bifida and the Impact of Prenatal Diagnosis —United States, 1985–1994

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*Abbreviations

ATSDR	Agency for Toxic Substances and Disease Registry
CIO	Centers/Institute/Offices
EPO	Epidemiology Program Office
IHPO	International Health Program Office
NCCDPHP	National Center for Chronic Disease Prevention and Health Promotion
NCEH	National Center for Environmental Health
NCEHIC	National Center for Environmental Health and Injury Control
NCID	National Center for Infectious Diseases
NCIPC	National Center for Injury Prevention and Control
NCPS	National Center for Prevention Services
NIOSH	National Institute for Occupational Safety and Health

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Surveillance for Anencephaly and Spina Bifida and the Impact of Prenatal Diagnosis—United States, 1985–1994

Janet D. Cragan, M.D.
Helen E. Roberts, M.D.
Larry D. Edmonds, M.S.P.H.
Muin J. Khoury, M.D., Ph.D.
Division of Birth Defects and Developmental Disabilities
National Center for Environmental Health
Russell S. Kirby, Ph.D., M.S.
Arkansas Reproductive Health Monitoring System
Gary M. Shaw, Dr.P.H.
Ellen M. Velie, M.P.H.
California Birth Defects Monitoring Program
Ruth D. Merz, M.S.
Mathias B. Forrester, B.S.
Hawaii Birth Defects Monitoring Program
Roger A. Williamson, M.D.
Diane S. Krishnamurti, M.S.
Iowa Birth Defects Registry and the University of Iowa
Roger E. Stevenson, M.D.
Jane H. Dean, R.N.
Greenwood Genetic Center

Abstract

Problem/Condition: The reported prevalence of anencephaly and spina bifida in the United States has steadily declined since the late 1960s. During this time, the ability to diagnose these defects prenatally has progressed rapidly. Many U.S. birth defects surveillance systems ascertain defects only among live-born infants or among infants and fetuses beyond a certain gestational age, thus excluding defects among pregnancies prenatally diagnosed as being affected by a neural tube defect (NTD) and electively terminated before the gestational age limit. The impact of prenatal diagnosis and subsequent pregnancy termination on the reported prevalence of anencephaly and spina bifida in the United States has not been well established. However, assessment of this impact is crucial to the use of surveillance data to monitor trends in the occurrence of NTDs and the effectiveness of interventions for these defects (e.g., increased consumption of folic acid).

Reporting Period: This report presents data from birth defects surveillance systems in six states over different time periods: Arkansas, 1985–1989; California, 1989–1991; Georgia, 1990–1991; Hawaii, 1988–1994; Iowa, 1985–1990; and South Carolina, 1992–1993.

Description of Systems: Population-based data about a) live-born and stillborn infants with anencephaly and spina bifida and b) pregnancies electively terminated after

prenatal diagnosis of these defects were analyzed from the Arkansas Reproductive Health Monitoring System; the California Birth Defects Monitoring Program; CDC's Metropolitan Atlanta Congenital Defects Program; the Iowa Birth Defects Registry, the University of Iowa, and the Iowa Department of Public Health; and the Greenwood Genetic Center in South Carolina. Data also were analyzed from the Hawaii Birth Defects Monitoring Program, which includes data for some women who were not residents of the state. The systems differed in the size and racial/ethnic composition of the populations studied, the surveillance methods used, the completeness of ascertainment, and the availability and utilization of prenatal testing and pregnancy termination.

Results and Interpretation: Among all pregnancies ascertained in which the infant or fetus had anencephaly or spina bifida, the percentages that were electively terminated ranged from 9% in Arkansas to 42% in Atlanta and Hawaii, with a corresponding increase in the adjusted prevalence of these defects compared with the prevalence at birth. In each system, pregnancies associated with anencephaly were terminated more frequently than were those associated with spina bifida. These data indicate that the impact of prenatal diagnosis and subsequent pregnancy termination on the prevalence at birth of anencephaly and spina bifida differs among geographic areas and populations. Comprehensive surveillance for these defects requires inclusion of pregnancies that are prenatally diagnosed and then terminated.

Actions Taken: CDC will use these data to promote the inclusion of prenatally diagnosed and terminated pregnancies in estimates of the prevalence of anencephaly and spina bifida generated by birth defects surveillance programs in the United States. Including such pregnancies is crucial to the ability of these programs to monitor trends accurately and to establish the effectiveness of interventions, including the use of folic acid, for these defects.

INTRODUCTION

In the early 1970s, an association was documented between elevated alpha-fetoprotein levels in women during early pregnancy and the presence of anencephaly or open spina bifida, which are congenital neural tube defects (NTDs) of the brain and spinal cord, respectively (*1,2*). Since then, the ability to diagnose NTDs prenatally has progressed rapidly, with increased utilization of and improved techniques for both maternal serum alpha-fetoprotein (MSAFP) screening and high-resolution ultrasonography. Because many birth defects surveillance systems in the United States were established before these prenatal techniques were widely used, they ascertain defects primarily from hospital records among live-born infants only or among infants and fetuses beyond a certain gestational age (*3*). Pregnancies that are prenatally diagnosed with NTDs and subsequently terminated in an outpatient setting or before the specified gestational age often are not included in U.S. birth defects surveillance data.

The reported prevalence at birth of NTDs in the United States has steadily declined since the late 1960s (*4*), before the use of prenatal diagnostic testing became widespread. An undetermined proportion of this decline may reflect the fact that birth defects surveillance systems do not include NTD-affected pregnancies that are electively terminated after prenatal diagnosis. Reports from other countries (e.g., England, France, and Scotland) estimate that, in some areas in the mid-1980s, at least 80% of pregnancies affected by anencephaly and 40% of those affected by spina bifida were

electively terminated (*5*). The only similar published estimate from the United States indicates that, in one area during 1990, the percentage of pregnancies affected by anencephaly that were electively terminated may have been even higher (*6*).

This report summarizes findings from birth defects surveillance systems in six states during the period 1985–1994 that were able to ascertain NTD-affected pregnancies which were prenatally diagnosed and then electively terminated. These findings estimate the reduction in the reported prevalence at birth of anencephaly and spina bifida in the United States resulting from prenatal diagnosis and subsequent pregnancy termination. The findings emphasize the importance of including prenatally diagnosed defects in NTD surveillance data.

METHODS

Population-based data about a) live-born and stillborn infants who had anencephaly and spina bifida* and b) pregnancies electively terminated after prenatal diagnosis of these defects were analyzed from five U.S. birth defects surveillance systems: the Arkansas Reproductive Health Monitoring System (ARHMS); the California Birth Defects Monitoring Program (CBDMP); CDC's Metropolitan Atlanta Congenital Defects Program (MACDP); the Iowa Birth Defects Registry, the University of Iowa, and the Iowa Department of Public Health; and the Greenwood Genetic Center (GGC) in South Carolina. Data were also analyzed from the Hawaii Birth Defects Monitoring Program (HBDMP), which includes data for some women who were not residents of the state. The years surveyed, the size of the populations studied, and the methods of ascertainment differed among the systems (Table 1). The case definitions of anencephaly and spina bifida also differed.

For the purpose of these analyses, all ascertained pregnancies in which the fetuses were prenatally diagnosed as having anencephaly or spina bifida and which were electively terminated were excluded from the calculations of prevalence at birth, regardless of the gestational age at termination. These estimates are based on the assumption that all NTD-affected pregnancies, had they not been terminated, would have been included in the prevalences of these defects at birth. The data were analyzed for each system by race/ethnicity because of previously reported race-specific differences in the prevalences at birth of these defects (*7*); race/ethnicity was either reported by the mother or obtained from patient records.

Arkansas: The ARHMS collected data from approximately two-thirds of the state's population born during the period 1985–1987 and from approximately half of the state's population born during the period 1988–1989. A case was defined as either a) anencephaly and/or spina bifida in a live-born or stillborn infant of ≥22 weeks' gestation born from 1985 through 1989 or b) a pregnancy electively terminated during this same time period after prenatal diagnosis of these defects, regardless of the gestational age of the fetus. Cases were ascertained by review of medical records from hospitals, genetic service clinics, and specialty clinics, as well as through passive reporting by schools and community agencies. Race/ethnicity was categorized as either "white" or "black."† Data from other racial and ethnic groups were not analyzed because of the limited number of cases among these groups. The population base from

*The data do not distinguish between open and closed spina bifida.
†Whether Hispanic infants were included in this category is unknown.

TABLE 1. Comparison of selected birth defects surveillance systems — United States

System	Years surveyed	Reporting area	Total no. of births*	Methods of ascertainment
Arkansas Reproductive Monitoring System	1985–1989	1985–1987: 67% of the state population; 1988–1989: 50% of the state population	108,519	Record review: hospitals, genetic and specialty health clinics; Passive reporting: schools, community agencies
California Birth Defects Monitoring Program	1989–1991†	All counties except Los Angeles, Ventura, and Riverside	708,129	Record review: hospitals, genetic clinics, and ultrasonography records
Metropolitan Atlanta Congenital Defects Program	1990–1991	Five-county metropolitan Atlanta area	77,022	Record review: hospitals, genetic laboratories, perinatal offices, and vital records
Hawaii Birth Defects Monitoring Program	1988–1994	Statewide	148,092	Record review: hospitals, laboratories, prenatal diagnostic centers, and vital records
Iowa Birth Defects Registry, the University of Iowa, and the Iowa Department of Public Health	1985–1990	Statewide	234,113	Data linkage: Birth Defects Registry, Prenatal Diagnosis Clinic, Fetal Diagnosis and Treatment Unit, and the MSAFP Screening Program
Greenwood Genetic Center (South Carolina)	1992–1993§	14 upstate counties	16,641	Record review: MSAFP and autopsy programs, obstetric offices Periodic hospital reports

* Includes live-born and stillborn infants ≥20 weeks' gestation in the California, Iowa, and Arkansas systems; live-born infants in the Atlanta system; live-born and stillborn infants who either were >20 weeks' gestation or weighed ≥ 350 grams at birth in the South Carolina system; and live-born and stillborn infants or fetuses of any gestational age in the Hawaii system.
† Data were collected for live-born and stillborn infants who were born from June 1, 1989, through May 31, 1991. Data were collected for pregnancies that were terminated from February 1, 1989, through January 31,1991.
§ Data were collected for pregnancies with an estimated date of delivery from October 1, 1992, through September 30, 1993.
MSAFP = Maternal serum alpha-fetoprotein

which defect prevalences were calculated consisted of 108,519 live-born and stillborn infants of ≥20 weeks' gestation.

California: The CBDMP collected data from all counties in the state except Los Angeles, Ventura, and Riverside. A case was defined as either a) anencephaly and/or spina bifida in a live-born or stillborn infant of ≥20 weeks' gestation born during the period June 1, 1989, through May 31, 1991, or b) a pregnancy electively terminated from February 1, 1989, through January 31, 1991, after prenatal diagnosis of these defects, regardless of the gestational age of the fetus. Cases were ascertained by review of medical and ultrasonography records at all hospitals and genetic clinics serving the population base. For this evaluation, race/ethnicity was categorized as either "white" or "other," with "other" including all racial and ethnic groups other than white. The population base from which defect prevalences were calculated consisted of 708,129 live-born and stillborn infants of ≥20 weeks' gestation.

Atlanta, Georgia: The MACDP collected data from the five-county metropolitan Atlanta area. A case was defined as either a) anencephaly and/or spina bifida in a live-born or stillborn infant of ≥20 weeks' gestation born during the period 1990–1991 or b) a pregnancy electively terminated after prenatal diagnosis of these defects, in which the fetus was of any gestational age but would have been at least 20 weeks' gestation during this same time period had the pregnancy not been terminated. Cases were ascertained by review of medical records from all hospitals, outpatient perinatal centers, and local genetic laboratories within the five-county metropolitan area, as well as from vital records. Race/ethnicity was categorized as either "white" or "black."* Data from other racial and ethnic groups were not analyzed because of the limited number of cases among these groups. The population base from which defect prevalences were calculated consisted of 77,022 live-born infants.

Hawaii: The HBDMP collected data from the entire state. A case was defined as either a) anencephaly and/or spina bifida in a live-born or stillborn infant or fetus of any gestational age born in Hawaii during the period 1988–1994 or b) a pregnancy electively terminated in Hawaii during this same period after prenatal diagnosis of these defects, regardless of the gestational age of the fetus. Because cases were included regardless of whether the mother resided in Hawaii, the HBDMP data were not strictly population based. Cases were ascertained by review of medical records from all birth and tertiary hospitals, laboratories, and prenatal diagnostic centers in the state, as well as from vital records. Race/ethnicity was categorized as "white" or Asian. Data from other racial and ethnic groups, including Hispanic, were not analyzed because of the limited number of cases among these groups. The population base from which defect prevalences were calculated consisted of 148,092 live-born and stillborn infants and fetuses of any gestational age.

Iowa: Statewide data were collected from the Iowa Birth Defects Registry, the Maternal Serum Alpha-Fetoprotein Screening Program (directed by the Iowa Department of Public Health), and the Prenatal Diagnosis Clinic and Fetal Diagnosis and Treatment Unit of the University of Iowa Hospitals and Clinics. A case was defined as either a) anencephaly and/or spina bifida not associated with other major malformations or clinical syndromes in a live-born or stillborn infant of ≥20 weeks' gestation born from 1985 through 1990 or b) a pregnancy electively terminated during this period after prenatal diagnosis of these defects, regardless of the gestational age of the fetus. The population base from which defect prevalences were calculated consisted of 234,113

*Whether Hispanic infants were included in this category is unknown.

live-born and stillborn infants of ≥20 weeks' gestation, approximately 96% of whom were white.*

South Carolina: The GGC collected data from 14 counties that comprise approximately 30% of the state's population. A case was defined as either a) a pregnancy in which the mother had an estimated date of delivery during the period October 1, 1992, through September 30, 1993, that resulted in a live-born or stillborn infant of any gestational age with anencephaly and/or spina bifida or b) a pregnancy in which the mother had an estimated date of delivery during this same time period and which was electively terminated after prenatal diagnosis of these defects, regardless of the gestational age of the fetus. Cases were ascertained by continuous monitoring of MSAFP screening programs, obstetric offices, and fetal/neonatal autopsy programs serving the population base, as well as by periodic monitoring of hospital medical record departments and neonatal intensive-care units. Race/ethnicity was categorized as either "white" or "other."* All but one of the NTD-affected infants and fetuses in the "other" category were black.* The population base from which defect prevalences were calculated consisted of 16,641 live-born and stillborn infants who either were ≥20 weeks' gestation or weighed ≥350 grams at birth.

RESULTS

Among all pregnancies ascertained in which the infant or fetus had anencephaly or spina bifida, from 9% in Arkansas to 42% in Atlanta and Hawaii were electively terminated, with a corresponding increase in the adjusted prevalence of these defects compared with the prevalence at birth (Table 2). In each system, pregnancies associated with anencephaly were terminated more frequently (range: 20% in Arkansas to 69% in Hawaii) than were those associated with spina bifida (range: 3% in Arkansas to 29% in California).†

The adjusted prevalences of anencephaly and spina bifida, both individually and combined, also differed among the systems. The prevalence in each category was highest in South Carolina and lowest in Hawaii. In every system, the adjusted prevalence of spina bifida was higher than that of anencephaly; however, the prevalence was only slightly higher in Atlanta.

Among all pregnancies ascertained in which the infant or fetus had anencephaly or spina bifida, the percentages that were electively terminated were available for each year of surveillance for Arkansas, Hawaii, and Iowa (Table 3). In Arkansas, this percentage more than tripled from 1985 (7%) to 1989 (23%); in Iowa, the percentage doubled from 1985 (13%) to 1990 (27%); in Hawaii, the percentage varied over the years without a discernible trend (range: 30% to 67%).§ However, in Hawaii, the adjusted prevalence of these defects almost doubled from the earlier years of surveillance

*Whether Hispanic infants were included in this category is unknown.

†It is unknown whether four of the NTD-affected pregnancies in Hawaii (two with anencephaly, two with spina bifida) were electively terminated. If all four pregnancies were prenatally diagnosed and subsequently terminated, 73% of the pregnancies with anencephaly, 26% of those with spina bifida, and 46% of the total NTD-affected pregnancies from Hawaii would have been electively terminated.

§It is unknown whether four of the NTD-affected pregnancies in Hawaii (two with anencephaly, two with spina bifida) were electively terminated. If all four of these pregnancies were prenatally diagnosed and subsequently terminated, the range of NTD-affected pregnancies that were electively terminated in Hawaii would have been 33% to 67%.

TABLE 2. Prevalence at birth of anencephaly and spina bifida and adjusted prevalence after prenatal diagnosis and elective termination,* by birth defect and system — United States

Birth defect/ System	Prevalence at birth†			No. of terminated pregnancies	Adjusted prevalence§			Percentage of pregnancies terminated¶
	No.	Rate	(95% CI)		No.	Rate	(95% CI)	
Anencephaly								
ARHMS	32	0.29	(0.20–0.42)	8	40	0.37	(0.26–0.55)	20
CBDMP	143	0.20	(0.17–0.24)	142	285	0.40	(0.36–0.45)	50
MACDP	15	0.19	(0.11–0.32)	22	37	0.48	(0.34–0.66)	59
HBDMP	12	0.08	(0.04–0.14)	31	45**	0.30	(0.22–0.41)	69
Iowa Birth Defects Registry††	62	0.27	(0.20–0.34)	20	82	0.35	(0.28–0.43)	24
GGC	4	0.24	(0.06–0.62)	6	10	0.60	(0.29–1.11)	60
Spina bifida								
ARHMS	70	0.65	(0.50–0.82)	2	72	0.66	(0.52–0.84)	3
CBDMP	250	0.35	(0.31–0.40)	103	353	0.50	(0.45–0.55)	29
MACDP	29	0.38	(0.25–0.54)	10	39	0.51	(0.36–0.69)	26
HBDMP	45	0.30	(0.22–0.41)	14	61**	0.41	(0.32–0.53)	23
Iowa Birth Defects Registry	104	0.44	(0.36–0.54)	25	129	0.55	(0.46–0.65)	19
GGC	12	0.72	(0.37–1.26)	4	16	0.96	(0.55–1.56)	25
Total								
ARHMS	102	0.94	(0.76–1.14)	10	112	1.03	(0.85–1.24)	9
CBDMP	393	0.55	(0.50–0.61)	245	638	0.90	(0.83–0.97)	38
MACDP	44	0.57	(0.42–0.77)	32	76	0.99	(0.78–1.23)	42
HBDMP	57	0.38	(0.29–0.50)	45	106**	0.72	(0.59–0.87)	42
Iowa Birth Defects Registry	166	0.71	(0.61–0.83)	45	211	0.90	(0.78–1.07)	21
GGC	16	0.96	(0.55–1.56)	10	26	1.56	(1.02–2.29)	38

*Both prevalences are per 1,000 live-born and stillborn infants ≥20 weeks' gestation in the California, Iowa, and Arkansas systems; per 1,000 live-born infants in the Atlanta system; per 1,000 live-born and stillborn infants who were >20 weeks' gestation or had a birth weight of ≥ 350 grams in the South Carolina system; and per 1,000 live-born and stillborn infants or fetuses of any gestational age in the Hawaii system.

†Includes live-born and stillborn infants ≥20 weeks' gestation in the California, Iowa, South Carolina, and Atlanta systems; live-born and stillborn infants ≥22 weeks' gestation in the Arkansas system; and live-born and stillborn infants and fetuses of any gestational age in the Hawaii system.

§Includes a) pregnancies that were electively terminated and b) infants who were included in the calculations for birth prevalence.

¶Percentage of pregnancies included in the calculations for adjusted prevalence that were electively terminated.

**It is unknown whether four of the prenatally diagnosed pregnancies in Hawaii (two with anencephaly, two with spina bifida) were electively terminated. If all four pregnancies had been prenatally diagnosed and subsequently terminated, 73% of the pregnancies with anencephaly, 26% of those with spina bifida, and 46% of the total NTD-affected pregnancies from Hawaii would have been electively terminated.

††The Iowa Birth Defects Registry includes data from the University of Iowa and the Iowa Department of Public Health.

ARHMS = Arkansas Reproductive Health Monitoring System
CBDMP = California Birth Defects Monitoring Program
MACDP = Metropolitan Atlanta Congenital Defects Program
HBDMP = Hawaii Birth Defects Monitoring Program
GGC = Greenwood Genetic Center (South Carolina)
95% CI = 95% confidence intervals

TABLE 3. Prevalence at birth of anencephaly and spina bifida and adjusted prevalence after prenatal diagnosis and elective termination,* by year, selected birth defects surveillance systems — United States

System/Year	Prevalence at birth†			No. of terminated pregnancies	Adjusted prevalence§			Percentage of pregnancies terminated¶
	No.	Rate	(95% CI)		No.	Rate	(95% CI)	
ARHMS								
1985	28	1.14	(0.76–1.65)	2	30	1.23	(0.83–1.75)	7
1986	26	1.08	(0.71–1.59)	0	26	1.08	(0.71–1.59)	—
1987	19	0.91	(0.55–1.43)	1	20	0.96	(0.59–1.48)	5
1988	12	0.62	(0.32–1.08)	2	14	0.72	(0.39–1.21)	14
1989	17	0.86	(0.50–1.37)	5	22	1.11	(0.70–1.68)	23
HBDMP								
1988	7	0.34	(0.14–0.71)	4	11	0.54	(0.27–1.00)	36
1989	8	0.38	(0.12–0.89)	4	12	0.57	(0.30–1.00)	33
1990	5	0.22	(0.07–0.52)	6	12**	0.54	(0.28–0.94)	50
1991	2	0.09	(0.01–0.34)	4	6	0.28	(0.10–0.61)	67
1992	12	0.56	(0.29–0.98)	10	22	1.00	(0.65–1.56)	45
1993	13	0.62	(0.33–1.06)	6	20**	0.97	(0.59–1.50)	30
1994	10	0.49	(0.23–0.89)	11	23**	1.11	(0.61–1.87)	48
Iowa††								
1985	34	0.83	(0.57–1.15)	5	39	0.95	(0.67–1.29)	13
1986	29	0.75	(0.50–1.07)	4	33	0.85	(0.59–1.20)	12
1987	29	0.77	(0.51–1.10)	6	35	0.92	(0.64–1.29)	17
1988	21	0.55	(0.34–0.84)	7	28	0.74	(0.49–1.06)	25

* Both prevalences are per 1,000 live-born and stillborn infants ≥20 weeks' gestation in the Arkansas and Iowa systems, and per 1,000 live-born and stillborn infants or fetuses of any gestational age in the Hawaii system.

† Includes live-born and stillborn infants ≥20 weeks' gestation in the Iowa system, live-born and stillborn infants ≥22 weeks' gestation in the Arkansas system, and live-born and stillborn infants and fetuses of any gestational age in the Hawaii system.

§ Includes a) pregnancies that were electively terminated and b) infants who were included in the calculations for prevalence at birth.

¶ Percentage of pregnancies included in the calculations for adjusted prevalence that were electively terminated.

** It is unknown whether four of the prenatally diagnosed pregnancies in Hawaii (two with anencephaly, two with spina bifida) were electively terminated. If all four pregnancies had been prenatally diagnosed and sibsequently terminated, 58% of the NTD-affected pregnancies from 1990, 35% of those from 1993, and 57% of those from 1994 in Hawaii would have been electively terminated.

†† The Iowa Birth Defects Registry includes data from the University of Iowa and the Iowa Department of Public Health.

ARHMS = Arkansas Reproductive Health Monitoring System
HBDMP = Hawaii Birth Defects Monitoring Program
95% CI = 95% confidence intervals

(1988–1991, range: 0.28 to 0.57 per 1,000) to the later years (1992–1994, range: 0.97 to 1.11).

The effect of prenatal diagnosis and subsequent termination on the prevalence of anencephaly and spina bifida was compared among racial groups in Arkansas, Atlanta, and Hawaii (Table 4). The prevalences among racial groups in the other systems were not compared because of the limited number of cases among many of those groups. In both Arkansas and Atlanta, the prevalence at birth and adjusted prevalence of anencephaly and spina bifida were higher among white women than among black women. In Atlanta, a higher percentage of ascertained NTD-affected pregnancies was electively terminated among white women. In Arkansas, although a limited number of pregnancies among black women was ascertained, a higher percentage of those affected by anencephaly was electively terminated compared with those among white women.

In Hawaii, the adjusted prevalence of anencephaly and its prevalence at birth were similar among white women and Asian women; however, the prevalence at birth and adjusted prevalence of spina bifida were higher among white women. For both defects, the percentage of ascertained pregnancies that were subsequently terminated was higher among Asian women.

DISCUSSION

These data provide the first multistate, population-based estimate of the impact of prenatal diagnosis and subsequent pregnancy termination on the prevalence at birth of anencephaly and spina bifida in the United States. In some areas, the prevalence at birth of anencephaly was reduced by approximately 60%–70% and that of spina bifida by approximately 20%–30%.

Among the six systems, the percentage of NTD-affected pregnancies that were prenatally diagnosed and subsequently terminated varied widely. This variation may reflect differences in surveillance methods, completeness of ascertainment, availability and utilization of prenatal diagnostic testing, and acceptance of elective pregnancy termination. Comprehensive ascertainment of NTD-affected pregnancies that are subsequently terminated can be particularly difficult and variable among systems. In all six systems, some women who had NTD-affected pregnancies that were prenatally diagnosed and then terminated without being referred to a specialty center participating in the surveillance system might not have been included in the estimates of adjusted defect prevalence. As a result, both the number of prenatally diagnosed and terminated pregnancies and the estimated percentages of all NTD-affected pregnancies that were electively terminated reported by each system would have been decreased.

Effects of Different Methods for Ascertaining Cases

Statewide MSAFP screening programs, which can facilitate the prenatal diagnosis of fetuses with NTDs, are maintained in both California and Iowa. However, some MSAFP specimens might not have been submitted through these programs, thus lowering the number of prenatally diagnosed pregnancies ascertained by these systems. In addition, infants and fetuses with anencephaly and spina bifida associated with other major malformations were excluded from the case definition in Iowa. This exclusion might have lowered the ascertained prevalences of these defects at birth and the

TABLE 4. Prevalence at birth of anencephaly and spina bifida and adjusted prevalence after prenatal diagnosis and elective termination,* by race–selected birth defects surveillance systems — United States

System/Race	Prevalence at birth†			No. of terminated pregnancies	Adjusted prevalence§			Percentage of pregnancies terminated¶
	No.	Rate	(95% CI)		No.	Rate	(95% CI)	
Anencephaly								
ARHMS								
White	27	0.32	(0.21–0.47)	6	33	0.39	(0.27–0.55)	18
Black	3	0.13	(0.03–0.39)	2	5	0.22	(0.07–0.51)	40
MACDP								
White	9	0.21	(0.10–0.40)	16	25	0.58	(0.37–0.86)	64
Black	5	0.16	(0.05–0.37)	3	8	0.25	(0.11–0.50)	38
HBDMP								
White	3	0.08	(0.02–0.24)	6	9	0.24	(0.11–0.46)	67
Asian	6	0.07	(0.02–1.46)	17	23	0.27	(0.16–0.38)	74
Spina bifida								
ARHMS								
White	57	0.68	(0.51–0.88)	2	59	0.70	(0.53–0.97)	3
Black	12	0.53	(0.27–0.92)	0	12	0.53	(0.27–0.92)	—
MACDP								
White	18	0.42	(0.25–0.66)	8	26	0.60	(0.39–0.88)	31
Black	11	0.35	(0.17–0.62)	2	13	0.41	(0.22–0.70)	15
HBDMP								
White	16	0.43	(0.25–0.70)	2	18	0.48	(0.29–0.76)	11
Asian	27	0.30	(0.20–0.44)	7	34	0.39	(0.26–0.53)	21
Total								
ARHMS								
White	84	1.00	(0.79–1.23)	8	92	1.09	(0.88–1.34)	9
Black	15	0.66	(0.34–1.09)	2	17	0.75	(0.44–1.20)	12
MACDP								
White	27	0.63	(0.41–0.91)	24	51	1.18	(0.88–1.55)	47
Black	16	0.51	(0.29–0.82)	5	21	0.66	(0.41–1.01)	24
HBDMP								
White	19	0.51	(0.31–0.80)	8	27	0.72	(0.48–1.10)	30
Asian	33	0.37	(0.25–0.52)	24	57	0.63	(0.48–0.82)	42

* Both prevalences are per 1,000 live-born and stillborn infants ≥20 weeks' gestation in the Arkansas system; per 1,000 live-born infants in the Atlanta system; and per 1,000 live-born and stillborn infants or fetuses of any gestational age in the Hawaii system.
† Includes live-born and stillborn infants ≥22 weeks' gestation in the Arkansas system; live-born and stillborn infants ≥ 20 weeks' gestation in the Atlanta system; and live-born and stillborn infants and fetuses of any gestational age in the Hawaii system.
§ Includes a) pregnancies that were electively terminated and b) infants who were included in the calculations for prevalence at birth.
¶ Percentage of pregnancies included in the calculations for adjusted prevalence that were electively terminated.
ARHMS = Arkansas Reproductive Health Monitoring System
MACDP = Metropolitan Atlanta Congenital Defects Program
HBDMP = Hawaii Birth Defects Monitoring Program
95% CI = 95% confidence intervals

estimated percentages of all NTD-affected pregnancies that were electively terminated in Iowa compared with other systems.

Because MSAFP screening was not widely available in local health departments in Arkansas until the late 1980s, the estimated percentage of all NTD-affected pregnancies that were electively terminated reported by Arkansas was low. The availability of screening may partially account for the increase in this percentage during 1988 and 1989. Decreased funding for the surveillance program may also have affected case-finding in Arkansas during 1988 and 1989. In Atlanta, an MSAFP screening program has not been established; therefore, prenatally diagnosed pregnancies were ascertained primarily through review of ultrasonography records and amniotic fluid alpha-fetoprotein results.

Similarly, in Hawaii, cases were ascertained primarily through record review at prenatal diagnostic centers and birth hospitals. Because these records did not always indicate the final outcome of the pregnancy, it is unknown whether four of the NTD-affected pregnancies in Hawaii (two with anencephaly and two with spina bifida) were electively terminated. If all four of these pregnancies were terminated after prenatal diagnosis, the estimated percentage of NTD-affected pregnancies that were terminated in Hawaii would have been higher (Tables 2, 3). In addition, ascertainment of prenatally diagnosed pregnancies that were electively terminated was begun for all years of the surveillance in 1994 on a retrospective basis. The comprehensiveness of case ascertainment among these pregnancies might therefore be lower than that among live-born and stillborn infants and fetuses.

In contrast, the South Carolina system was based on direct contact with both local MSAFP programs and individual obstetricians and pathologists. This method might have provided more comprehensive ascertainment of prenatally diagnosed pregnancies in South Carolina compared with the other systems and might have contributed to the increased prevalences of these defects reported by South Carolina. However, the methods used to identify live-born and stillborn infants with anencephaly and spina bifida from hospital records in South Carolina may have resulted in less complete ascertainment of these cases than in other systems, resulting in a lowering of the estimated prevalence at birth in that state.

This analysis could not determine whether the decreased prevalences reported from Hawaii and the increased prevalences reported from South Carolina were true or whether they resulted from the differences in surveillance methods among the systems. In addition, the increased prevalences reported from South Carolina may also have resulted from the limited total number of NTD-affected pregnancies ascertained by that system.

Factors Affecting Calculations of NTD Prevalence at Birth

With regard to the data for each year of surveillance, the increased percentage of all ascertained NTD-affected pregnancies that were electively terminated in the later years in the Arkansas and Iowa systems may reflect an increase in the availability and utilization of prenatal diagnostic procedures, an increased ability of physicians to diagnose NTDs prenatally, an increase in the referral of pregnancies suspected to be affected with NTDs to subspecialists, and improved case ascertainment. The lack of a similar trend in these percentages over time in the Hawaii system may reflect the relatively later years of surveillance reported by that system and a more uniform pattern of prenatal care practices compared with the other systems. The reason for the

increase in prevalence at birth and adjusted prevalence in Hawaii for 1992–1994 compared with 1988–1991 is unclear. Neither surveillance techniques nor case ascertainment methods changed during those years. Continued surveillance should clarify whether this increase in prevalence represents a persistent trend.

The comparison of data among racial groups from Arkansas, Atlanta, and Hawaii suggests an increased adjusted prevalence of anencephaly and spina bifida among white women compared with black women and an increased adjusted prevalence of spina bifida among white women compared with Asian women. These race-specific differences in the percentage of pregnancies terminated may have resulted from a) differences in availability and utilization of prenatal procedures, including elective termination, or b) the limited number of NTD-affected pregnancies among some racial groups ascertained by these systems.

In the six surveillance systems included in this report, all NTD-affected pregnancies that were subsequently terminated were excluded from the calculations of prevalence at birth of anencephaly and spina bifida. However, in some U.S. birth defects surveillance systems, only NTD-affected pregnancies that were electively terminated beyond a specific gestational age are included in the calculations of prevalence at birth. In addition, some surveillance systems may include infants with encephalocele and other NTDs in their case ascertainment. As a consequence, the reduction in the estimates of prevalence at birth resulting from prenatal diagnosis and subsequent termination calculated from some surveillance data may be smaller than reflected in this report. For example, if the MACDP data had included only NTD-affected pregnancies that were electively terminated at ≥20 weeks and if infants with encephalocele had been included in the MACDP case definition, the reduction in the prevalences at birth of NTDs attributable to prenatal diagnosis and elective termination would have been approximately 30% (*8*), not the 42% cited in this report.

CONCLUSIONS

The findings in this report indicate that the impact of prenatal diagnosis and subsequent pregnancy termination on surveillance for anencephaly and spina bifida can differ considerably among geographic areas, among populations, and over time. This variation underscores the necessity of monitoring this impact for each population—or subgroup of a population—studied. The findings also demonstrate the considerable magnitude of the reduction in prevalence at birth of these defects resulting from the widespread use of prenatal diagnostic techniques. Comprehensive surveillance for NTDs can no longer be conducted without ascertaining pregnancies that are prenatally diagnosed and then electively terminated.

Such comprehensive surveillance can play a key role in evaluating the effectiveness of preventive measures for NTDs. Improving the dietary level of folic acid (a B vitamin) has been demonstrated to prevent the occurrence of many NTDs (*9,10*). This finding represents a historic opportunity for the prevention of birth defects. In 1991 and 1992, CDC published recommendations for the use of folic acid to prevent NTDs (*11,12*). As these recommendations are implemented and the use of folic acid becomes more widespread, its effect on the prevalence of NTD-affected pregnancies must be closely and accurately monitored. If surveillance is to be used to monitor the effectiveness of this prevention and any resultant decline in the prevalence of NTDs attributable to folic acid use, pregnancies that are electively terminated after prenatal

diagnosis of an NTD must be included in NTD surveillance. Otherwise, evaluation of a reduction in the prevalence of NTDs attributable to folic acid cannot be distinguished from the decrease resulting from prenatal diagnosis and elective termination.

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State and Territorial Epidemiologists and Laboratory Directors

State and Territorial Epidemiologists and Laboratory Directors are acknowledged for their contributions to *CDC Surveillance Summaries*. The epidemiologists listed below were in the positions shown as of June 1995, and the laboratory directors listed below were in the positions shown as of June 1995.

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